

WHAT IS CLAIMED IS:

- 1                   1.     An isolated nucleic acid comprising a sequence selected from the  
2     group consisting of SEQ ID NO:9, SEQ ID NO: 10, SEQ ID NO: 11, and SEQ ID NO:12.
- 1                   2.     The isolated nucleic acid of Claim 1, wherein said nucleic acid is  
2     deoxyribonucleic acid.
- 1                   3.     An isolated nucleic acid, wherein said nucleic acid is the complement  
2     of the nucleic acid of Claim 1.
- 1                   4.     A vector comprising the nucleic acid of Claim 1.
- 1                   5.     A host cell comprising the vector of Claim 4.
- 1                   6.     An isolated protein encoded by the nucleic acid sequence set forth in  
2     SEQ ID NO:9.
- 1                   7.     A method for detecting mutations in *Rab38* comprising the steps of:  
2                   a)     amplifying at least a portion of *Rab38* from genomic DNA to  
3     yield a *Rab38* amplification product;  
4                   b)     purifying said *Rab38* amplification product; and  
5                   c)     sequencing said *Rab38* amplification product.
- 1                   8.     The method of Claim 7, wherein said amplifying is accomplished  
2     using a polymerase chain reaction.
- 1                   9.     The method of Claim 7, wherein said at least a portion of *Rab38* is  
2     selected from the group consisting of at least one *Rab38* exon, at least one *Rab38* intron, the  
3     *Rab38* 5' untranslated sequence, and the *Rab38* 3' untranslated sequence.
- 1                   10.    The method of Claim 9, wherein said at least one *Rab38* exon is  
2     selected from the group consisting of *Rab38* exon 1, *Rab38* exon 2, and *Rab38* exon 3.
- 1                   11.    The method of Claim 7, wherein said genomic DNA is mammalian  
2     genomic DNA.
- 1                   12.    The method of Claim 7, wherein said purifying is accomplished using  
2     size selection.

1                   13.     A method for detecting mutations in *Rab38* comprising the steps of:  
2                   a)     amplifying at least a portion of *Rab38* from genomic DNA to  
3     yield a *Rab38* amplification product;  
4                   b)     digesting said *Rab38* amplification product to yield a digested  
5     *Rab38* amplification product; and  
6                   c)     electrophoresing said digested *Rab38* amplification product.

1                   14.     The method of Claim 13, wherein said amplifying is accomplished  
2     using a polymerase chain reaction.

1                   15.     The method of Claim 13, wherein said at least a portion of *Rab38* is  
2     selected from the group consisting of at least one *Rab38* exon, at least one *Rab38* intron, the  
3     *Rab38* 5' untranslated sequence, and the *Rab38* 3' untranslated sequence.

1                   16.     The method of Claim 15, wherein said at least one *Rab38* exon is  
2     selected from the group consisting of *Rab38* exon 1, *Rab38* exon 2, and *Rab38* exon 3.

1                   17.     The method of Claim 13, wherein said genomic DNA is mammalian  
2     genomic DNA.

1                   18.     A method for screening for biologically active agents to modulate  
2     RAB38 activity, comprising the steps of:

3                   a)     providing:  
4                   i)     melanocytes comprising RAB38 activity, and  
5                   ii)    a candidate agent; and  
6                   b)     exposing said melanocytes to said candidate agent to yield  
7     treated melanocytes; and  
8                   c)     measuring the modulation of said RAB38 activity of said  
9     treated melanocytes by said candidate agent.

1                   19.     The method of Claim 18, wherein said RAB38 activity comprises  
2     GTPase activity.

1                   20.     The method of Claim 18, wherein said RAB38 activity comprises GTP  
2     binding activity.

- 1                   21.    The method of Claim 18, wherein said RAB38 activity comprises GDP  
2 release.
- 1                   22.    The method of Claim 18, wherein said RAB38 activity comprises  
2 TYRP1 trafficking to melanosomes.
- 1                   23.    The method of Claim 18, wherein said RAB38 activity comprises  
2 RAB38 trafficking to melanosomes.
- 1                   24.    A kit for screening for biologically active agents that modulate RAB38  
2 activity, comprising: a) plurality of melanocytes comprising RAB38 activity, wherein said  
3 melanocytes are provided within a container, and b) instructions for determination of RAB38  
4 activity in said melanocytes.
- 1                   25.    The kit of Claim 24, further comprising means to analyze RAB38  
2 activity.
- 1                   26.    The kit of Claim 25, wherein said means to analyze RAB38 activity  
2 comprises an assay to assess GTPase activity.
- 1                   27.    The kit of Claim 25, wherein said means to analyze RAB38 activity  
2 comprises an assay to assess GTP binding activity.
- 1                   28.    The kit of Claim 25, wherein said means to analyze RAB38 activity  
2 comprises an assay to assess GDP release.
- 1                   29.    The kit of Claim 25, wherein said means to analyze RAB38 activity  
2 comprises an assay to assess TYRP 1 trafficking to melanosomes.
- 1                   30.    The kit of Claim 25, wherein said means to analyze RAB38 activity  
2 comprises an assay to assess RAB38 trafficking to melanosomes.
- 1                   31.    A kit for detection of mutations in RAB38 comprising at least two  
2 primer sequences suitable for amplification of at least a portion of RAB38, and instructions  
3 for utilizing said kit.

1                   32.     The kit of Claim 31, wherein said primer sequences are selected from  
2 the group consisting of SEQ ID NO:17, SEQ ID NO:18, SEQ ID NO:19, SEQ ID NO:20,  
3 SEQ ID NO:21, and SEQ ID NO:22.

1                   33.     The kit of Claim 31, wherein said kit is suitable for use in the  
2 polymerase chain reaction.

1                   34.     The kit of Claim 31, further comprising reagents for digesting nucleic  
2 acid.

1                   35.     A kit for diagnosing defects in melanosome function, comprising  
2 melanocytes comprising *RAB38* and instructions for assessing defects in melanosome  
3 function.

1                   36.     The kit of Claim 35, further comprising means to analyze *RAB38*  
2 activity.

1                   37.     The kit of Claim 36, wherein said means to analyze *RAB38* activity  
2 comprises an assay to assess GTPase activity.

1                   38.     The kit of Claim 36, wherein said means to analyze *RAB38* activity  
2 comprises an assay to assess GTP binding activity.

1                   39.     The kit of Claim 36, wherein said means to analyze *RAB38* activity  
2 comprises an assay to assess GDP release.

1                   40.     The kit of Claim 36, wherein said means to analyze *RAB38* activity  
2 comprises an assay to assess TYRP1 trafficking to melanosomes.

1                   41.     The kit of Claim 36, wherein said means to analyze *RAB38* activity  
2 comprises an assay to assess *RAB38* trafficking to melanosomes.

1                   42.     A composition for modulating pigmentation of melanocytes,  
2 comprising a modulator of *RAB38* activity.

1                   43.     The composition of claim 42, wherein the modulator of *RAB38*  
2 activity is an enhancer of *RAB38* activity.

1                   44.     The composition of claim 42, wherein the modulator of RAB38  
2 activity is an inhibitor of RAB38 activity.

1                   45.     The composition of claim 44, wherein the inhibitor of RAB38 activity  
2 is selected from the group consisting of siRNA and intrabodies.

1                   46.     A method of modulating the pigmentation of a melanosome and  
2 changing skin color, the method comprising: contacting a skin surface with a modulator of  
3 RAB38 activity, thereby regulating the activity of RAB38.

1                   47.     The method of claim 46, wherein the modulator is an inhibitor of  
2 RAB38 activity that down-regulates RAB38 activity and lightens skin color.

1                   48.     The method of claim 46, wherein the modulator is an enhancer of  
2 RAB38 activity that up-regulates RAB38 activity and darkens skin color.